

## DISORDERS INCLUDED IN THE STEPONE® NEWBORN SCREENING PANEL

### DISORDERS DETECTED BY TANDEM MASS SPECTROMETRY

#### Acylcarnitine Profile

##### Fatty Acid Oxidation Disorders

Carnitine/Acylcarnitine Translocase Deficiency  
Carnitine Palmitoyl Transferase Deficiency Type I<sup>1</sup>  
3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency  
2,4-Dienoyl-CoA Reductase Deficiency<sup>1</sup>  
Medium Chain Acyl-CoA Dehydrogenase Deficiency  
Multiple Acyl-CoA Dehydrogenase Deficiency  
Neonatal Carnitine Palmitoyl Transferase Deficiency Type II  
Short Chain Acyl-CoA Dehydrogenase Deficiency  
Short Chain Hydroxy Acyl-CoA Dehydrogenase Deficiency  
Trifunctional Protein Deficiency  
Very Long Chain Acyl-CoA Dehydrogenase Deficiency

##### Organic Acid Disorders

3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency  
Glutaric Acidemia Type I  
Isobutyryl-CoA Dehydrogenase Deficiency  
Isovaleric Acidemia  
2-Methylbutyryl-CoA Dehydrogenase Deficiency  
3-Methylcrotonyl-CoA Carboxylase Deficiency  
3-Methylglutaconyl-CoA Hydratase Deficiency  
Methylmalonic Acidemias  
    Methylmalonyl-CoA Mutase Deficiency  
    Some Adenosylcobalamin Synthesis Defects  
    Maternal Vitamin B12 Deficiency  
Mitochondrial Acetoacetyl-CoA Thiolase Deficiency  
Propionic Acidemia  
Multiple CoA Carboxylase Deficiency  
Malonic Aciduria

##### X-Linked Adrenoleukodystrophy (X-ALD)

#### Amino Acid Profile

##### Amino Acid Disorders

Argininemia  
Argininosuccinic Aciduria  
5-Oxoprolinuria 1  
Carbamoylphosphate Synthetase Deficiency<sup>1</sup>  
Citrullinemia  
Homocystinuria  
Hypermethioninemia  
Hyperammonemia, Hyperornithinemia, Homocitrullinuria  
    Syndrome 1  
Hyperornithinemia with Gyral Atrophy<sup>1</sup>  
Maple Syrup Urine Disease  
Phenylketonuria  
    Classical/Hyperphenylalaninemia  
    Biopterin Cofactor Deficiencies  
Tyrosinemia  
    Transient Neonatal Tyrosinemia  
    Tyrosinemia Type I<sup>2</sup>  
    Tyrosinemia Type II  
    Tyrosinemia Type III

##### Other Observations

Hyperalimentation  
Liver Disease  
Medium Chain Triglyceride Oil Administration  
Presence of EDTA Anticoagulants in blood specimen  
Treatment with Benzoate, Pyvalic Acid, or Valproic Acid  
Carnitine Uptake Deficiency<sup>1</sup>

##### Lysosomal Storage Disorders (LSD)

Fabry, Gaucher, Krabbe Disease, Mucopolysaccharidosis  
Type I (MPS-I), Niemann-Pick, and Pompe

### DISORDERS DETECTED BY OTHER TECHNOLOGIES

Biotinidase Deficiency  
    Complete Deficiency  
    Partial Deficiency  
Congenital Adrenal Hyperplasia  
    Salt Wasting 21-Hydroxylase Deficiency  
    Simple Virilizing 21-Hydroxylase Deficiency  
Congenital Hypothyroidism  
Cystic Fibrosis (*not valid after 90 days of age*)<sup>\*</sup>

Galactosemia  
    Galactokinase Deficiency  
    Galactose-1-Phosphate Uridyltransferase Deficiency  
    Galactose-4-Epimerase Deficiency  
Glucose-6-Phosphate Dehydrogenase Deficiency  
Severe Combined Immunodeficiency (SCID)  
Sickle Cell and other Hemoglobinopathies  
    Hemoglobin S, S/C, S/Beta-Thalassemia, C, & E Diseases

The analyses conducted by PerkinElmer Genetics produce results that can be used by qualified physicians in the diagnosis of disorders described herein. Evidence of these conditions can be detected in the vast majority of affected individuals; however, due to genetic variability, age of patient at time of specimen collection, quality of specimen, health status of the patient, and other variables, such conditions may not be detected in all affected patients.

1 There is a lower probability of detection of this condition during the immediate newborn period.

2 Succinylacetone (SUAC) is the primary marker for Tyrosinemia Type 1.

\* For information on DNA Carrier Testing for children over 90 days of age, please call 866.463.6436.